

(a) amplifying two or more exons or partial exons of BRCA1 to produce amplified fragments, wherein the two or more exons or partial exons are amplified in a multiplex amplification reaction to produce multiplex amplification products for analysis;

(b) determining the sizes and amounts of amplified fragments in the multiplex amplification products and comparing the determined sizes or amounts to standard values for amplification of the same exons or partial exons of wild-type BRCA1 gene, wherein a difference in fragment size or amount is indicative of the presence of a mutation in the BRCA1 gene; and

(c) if no mutation is detected in the BRCA1 gene as a result of the determination of the sizes and amounts of the amplification fragments, determining the sequence of one or more exons of the BRCA1 gene.

54. The method according to claim 53, wherein at least exons 2, 11 and 20 of the BRCA1 gene, or portions thereof, are amplified in one multiplex amplification reaction.

55. The method according to claim 53, wherein exons 2, 5, 9 and 14 of the BRCA1 gene, or portions thereof, are amplified in one multiplex amplification reaction.

56. The method according to claim 53, wherein exons 3, 7 and 15 of the BRCA1 gene, or portions thereof, are amplified in one multiplex amplification reaction.

57. The method according to claim 53, wherein the sequence is determined by amplifying a selected one of the multiplex amplification products in an aliquot of the multiplex reaction mixture and then sequencing the amplified multiplex amplification products.

58. The method according to claim 57, wherein the amplification of the multiplex reaction mixture and the sequencing of the amplified multiplex reaction product are performed in a single vessel.

59. The method according to claim 57, wherein the multiplex reaction mixture is amplified by combining the multiplex reaction mixture with an amplification mixture containing two primers for the selected one of the multiplex reaction products, a mixture of dNTP's and a thermostable polymerase in a buffer suitable for amplification, and exposing the resulting combination to a first series of thermal cycles including at least an extension phase and a denaturation phase to produce an amplified mixture containing the amplified multiplex reaction product; adding a sequencing mixture comprising a chain terminating nucleoside triphosphate to the amplified mixture and exposing the resulting combination to a second series of thermal cycles including at least an extension phase and a denaturation phase to produce sequencing fragments; and evaluating the size of the sequencing fragments.

60. The method according to claim 59, wherein the thermostable polymerase is Thermo Sequenase®.

61. The method according to claim 59, wherein the sequencing mixture further comprises a fluorescently labeled sequencing primer.

62. The method according to claim 61, wherein the sequencing mixture further comprises a thermostable polymerase for sequencing which incorporates dideoxynucleosides into an extending oligonucleotide at a rate which is no less than about 0.4 times the rate of incorporation of deoxynucleosides in an amplification mixture.

63. The method according to claim 62, wherein the thermostable polymerase for sequencing is Thermo Sequenase®

64. A kit for testing a sample for mutations in the BRCA1 gene comprising a mixture of at least four oligonucleotide primers, said primers being selected to amplify at least